SLC25A1 gene

solute carrier family 25 member 1

Normal Function

The *SLC25A1* gene provides instructions for making a protein that is found in mitochondria, which are the energy-producing centers in cells. The SLC25A1 protein transports a molecule called citrate out of mitochondria in exchange for another molecule called malate, which is transported in. Within mitochondria, both citrate and malate participate in reactions that produce energy for cell activities. Citrate is transported out of mitochondria because it also has important functions in other parts of the cell. In particular, citrate is involved in the production of fats (lipids) and the regulation of glycolysis, which is another critical energy-producing process within cells.

Health Conditions Related to Genetic Changes

2-hydroxyglutaric aciduria

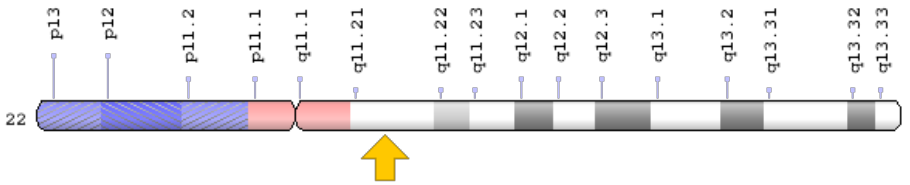
At least 12 mutations in the *SLC25A1* gene have been found to cause a form of 2-hydroxyglutaric aciduria called combined D,L-2-hydroxyglutaric aciduria (D,L-2-HGA). This condition causes severe brain abnormalities that become apparent in early infancy.

Each of the known *SLC25A1* gene mutations greatly reduces the function of the SLC25A1 protein. As a result, citrate and malate cannot be transported into and out of mitochondria, which disrupts energy production within cells. Through processes that are not fully understood, the lack of citrate and malate transport allows other compounds to build up abnormally within cells. These compounds include D-2-hydroxyglutarate and L-2-hydroxyglutarate, which at high levels can damage cells and lead to cell death. Brain cells appear to be the most vulnerable to the toxic effects of these compounds, which may explain why the signs and symptoms of D,L-2-HGA primarily involve the brain. Researchers suspect that an imbalance of other molecules, particularly citrate, also contributes to the severe signs and symptoms of combined D,L-2-HGA

Chromosomal Location

Cytogenetic Location: 22q11.21, which is the long (q) arm of chromosome 22 at position 11.21

Molecular Location: base pairs 19,175,575 to 19,178,863 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- citrate transport protein
- CTP
- D2L2AD
- SEA
- SLC20A3
- solute carrier family 20 (mitochondrial citrate transporter), member 3
- solute carrier family 25 (mitochondrial carrier; citrate transporter), member 1
- tricarboxylate carrier protein
- tricarboxylate transport protein, mitochondrial
- TXTP_HUMAN

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The Mitochondrion https://www.ncbi.nlm.nih.gov/books/NBK26894/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC25A1%5BTIAB%5D%29+OR+%28%28SLC20A3%5BTIAB%5D%29+OR+%28citrate+transport+protein%5BTIAB%5D%29+OR+%28tricarboxylate+carrier+protein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, CITRATE TRANSPORTER), MEMBER 1
  http://omim.org/entry/190315

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC25A1%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10979
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6576
- UniProt
  http://www.uniprot.org/uniprot/P53007

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